



**Foundation  
Fighting Blindness**

Driving research to save & restore sight

## **Usher Syndrome - Treatments**

While researchers are gaining new understandings about the precise genetic causes of Usher syndrome and the actual mechanisms of the disease, research has not yet found a way to halt the degeneration of the retina or to restore normal hearing. However, research is continuing in several different areas that can offer hope for people with Usher syndrome.

Possible treatments for Usher syndrome can be categorized by the two sensory organs that are affected:

### **THE EAR**

As we have seen, the hearing loss in Usher syndrome is due to an inner ear problem, which cannot currently be corrected with middle ear surgery. There is a possible medical device for hearing loss that may help some people with Usher syndrome.

A cochlear implant is a device that is surgically implanted in the mastoid bone and inner ear through an incision behind the ear. The device consists of a miniaturized magnetic receiver coil and an array of stimulating rings. The receiver is connected to the rings and embedded under the skin behind the ear. An external microphone and transmitter hook over the ear, attach magnetically to the receiver, and are connected to a battery-operated speech processing unit about the size of a beeper, which is worn at the waist or in a small backpack. The cochlear implant bypasses the non-functioning ciliated hair cells in the cochlea, taking over the job of converting sound waves into electrical energy and transmitting them to the auditory nerve. Cochlear implants work best in individuals who have some auditory memory; that is, they can recall what words and other sounds sound like. In recent years, they have been approved for implantation in young children who are profoundly deaf and must be taught the meaning of the sounds they are hearing.

Cochlear implants are controversial in some circles, and some deaf advocates argue that the procedure is still too experimental to use in children. They believe that there are better ways for people with a hearing impairment to function in the hearing world. Whether or not to get a cochlear implant is a very personal decision that people with Usher syndrome (or their parents, in the case of young children) must make based on a number of factors including the specifics of their hearing loss.

## **THE EYE**

Scientists have been very encouraged by the results of a six-year clinical study reported in June 1993 concerning vitamin A palmitate and RP. Researchers wanted to determine if increasing the amount of vitamin A palmitate in the diet could slow the progression of retinal degenerations. A carefully designed study showed that taking vitamin A palmitate can slow the progression of retinal degeneration for some people who have RP or Usher syndrome type II.

Subjects in the study included people with several common forms of RP and Usher syndrome type II. Patients with Usher syndrome type I were not studied. Therefore, recommendations cannot be made for those who have Usher syndrome type I. Patients with a daily consumption of about 18,000 International Units (IU) of vitamin A (15,000 IU palmitate in dietary supplements and about 3,000 from their regular diet) were found to have a lower rate of retinal degeneration, as measured by ERG, than patients not taking these doses of vitamin A. Taking vitamin A palmitate did not completely stop retinal degeneration, but the researchers found a 20 percent slower average annual decline of remaining retinal function in people taking the supplement. They concluded that the slowing could mean additional years of useful vision for many people with RP. For example, a person starting the daily supplement at age 32 could expect to retain some useful vision until the age of 70, while a person not taking the supplement would lose useful vision by age 63.

However, there are some warnings that accompany this recommendation. There is no evidence that doses higher than 15,000 IU provide greater benefits, and doses over 25,000 IU daily may be toxic and cause side effects such as liver disease. While there are no reported instances of toxicity in healthy adults taking 15,000 IU of vitamin A palmitate daily, people taking the supplement are advised to have their blood levels of vitamin A measured and have a test of their liver function before they begin the regimen, and annual tests thereafter. Also, because of the potential for birth defects, women who are pregnant or planning to become pregnant are not advised to take vitamin A palmitate in this dosage. This recommendation is for adults; RP patients under the age of 18 were not evaluated and 15,000 IU of supplementary vitamin A palmitate is not recommended for children. Parents of children with Usher syndrome should consult with their eye care professional and pediatrician about therapeutic doses of vitamin A palmitate for children, based on age and body size.

It is important to keep these findings in perspective. Vitamin A palmitate will not cure RP. Taking vitamin A will not improve your vision. The degenerative process will continue, but possibly at a slower rate. People with very advanced RP were not included in this study, and if you have advanced RP you should consult with your eye care professional about the possible benefit of vitamin A palmitate for you.

The same study that found these encouraging results from taking vitamin A palmitate supplements also looked at the effect of vitamin E supplements on retinal degeneration. In the case of vitamin E, the opposite effect was found. People taking 400 IU daily of vitamin E were found to have a faster rate of retinal degeneration than those in the other groups. This led to the recommendation that people with RP should avoid high-dose vitamin E supplements. However, there is no evidence that normal dietary or small supplemental amounts of vitamin E have an adverse effect on the progression of RP.

It is important that people with RP fully understand the implications of the findings regarding vitamin A and retinal degeneration. For a more complete discussion than is possible to include in this booklet (including a source list for this non-prescription supplement in the palmitate form as recommended in the study), contact The Foundation for a brochure, Vitamin A Treatment for Retinitis Pigmentosa.